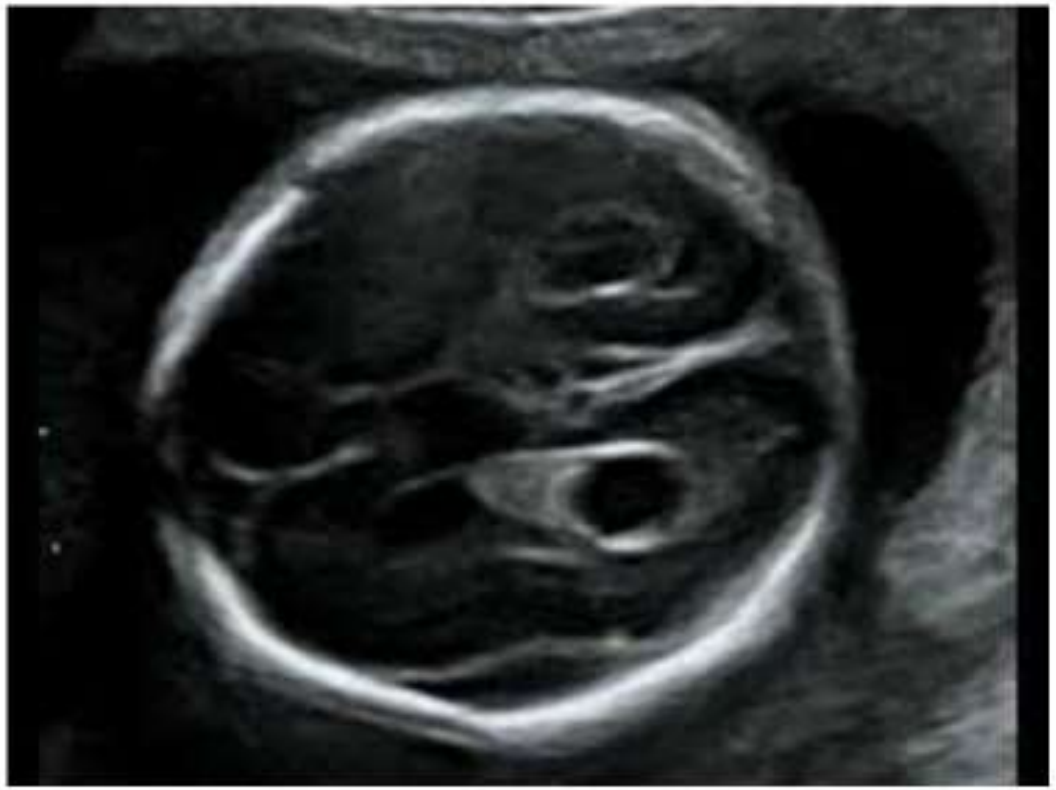
به نام خدا

Choroid plexus cysts



Choroid plexus cysts

Prevalence:

- 1 in 50 fetuses at 20 weeks' gestation.
- More than 90% resolve by 26 weeks.

Ultrasound diagnosis:

- Single or multiple cystic areas (>2 mm in diameter) in one or both choroid plexuses of the lateral cerebral ventricles.

Associated abnormalities:

- Associated with increased risk for trisomy 18 and possibly trisomy 21.

Investigations:

Detailed ultrasound examination for presence of other markers of trisomies 18 and 21. In the absence of other markers there is no need for invasive testing.

Follow up:

Follow-up should be standard.

Delivery:

Standard obstetric care and delivery.

Prognosis:

Good.

Recurrence risk:

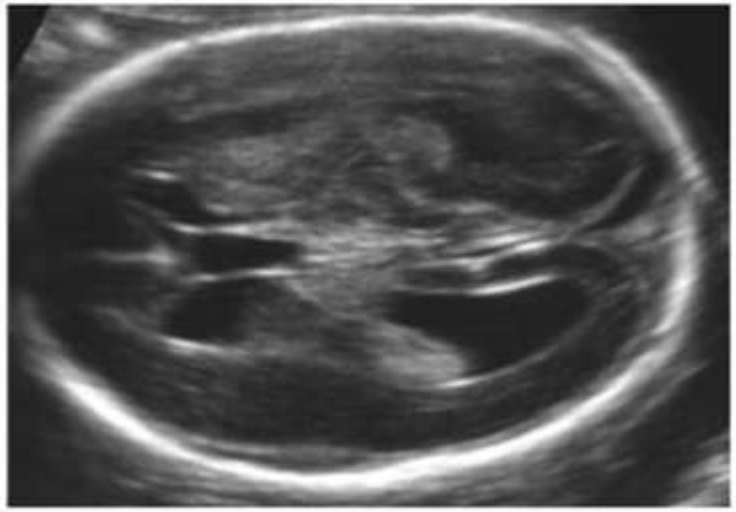
Isolated: no increased risk of recurrence.

Part of trisomies: 1%.

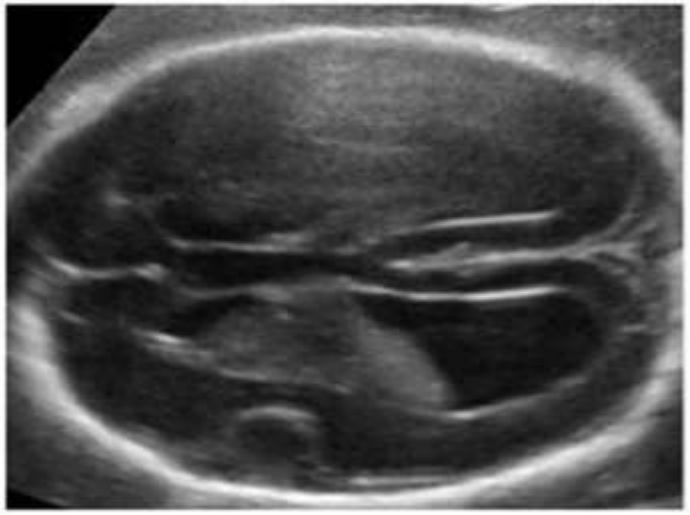
ventriculomegaly

isuog

The spectrum of mild cerebral lateral ventriculomegaly



10-12 mm



13-15 mm

Images by G. Pilu

Ventriculomegaly

Prevalence:

1 in 100 fetuses at 20 weeks' gestation.

1 in 1,000 births.

Ultrasound diagnosis:

Bilateral or unilateral dilation of the lateral cerebral ventricles observed in the standard transverse section of the brain.

Subdivided according to the diameter of the lateral ventricle into mild (10-12 mm), moderate (13-15 mm) and severe (>15 mm).

Associated abnormalities:

Chromosomal defects, mainly trisomies 21, 18 or 13, are found in 10% of cases. In isolated ventriculomegaly there is a 4-fold increase in risk for trisomy 21. Cerebral and non-cerebral defects and genetic syndromes are found in 50% of cases.

Investigations:

Detailed ultrasound examination, including neurosonography.

Invasive testing for karyotyping and array.

TORCH test for fetal infections.

Maternal blood testing for antiplatelet antibodies in cases with evidence of brain hemorrhage.

Follow up:

Ultrasound scans every 4 weeks to monitor the evolution of ventriculomegaly.

Delivery:

Standard obstetric care and delivery.

Cesarean section if the fetal head circumference is >40 cm.

Prognosis:

Isolated mild / moderate: neurodevelopmental delay in 10% of cases, this may not be higher than in the general population.

Isolated severe: 10 year survival 60%, severe mental handicap 50%.

Recurrence:

Isolated <1%. Increases to 5% if there is a history of affected fetus or sibling.

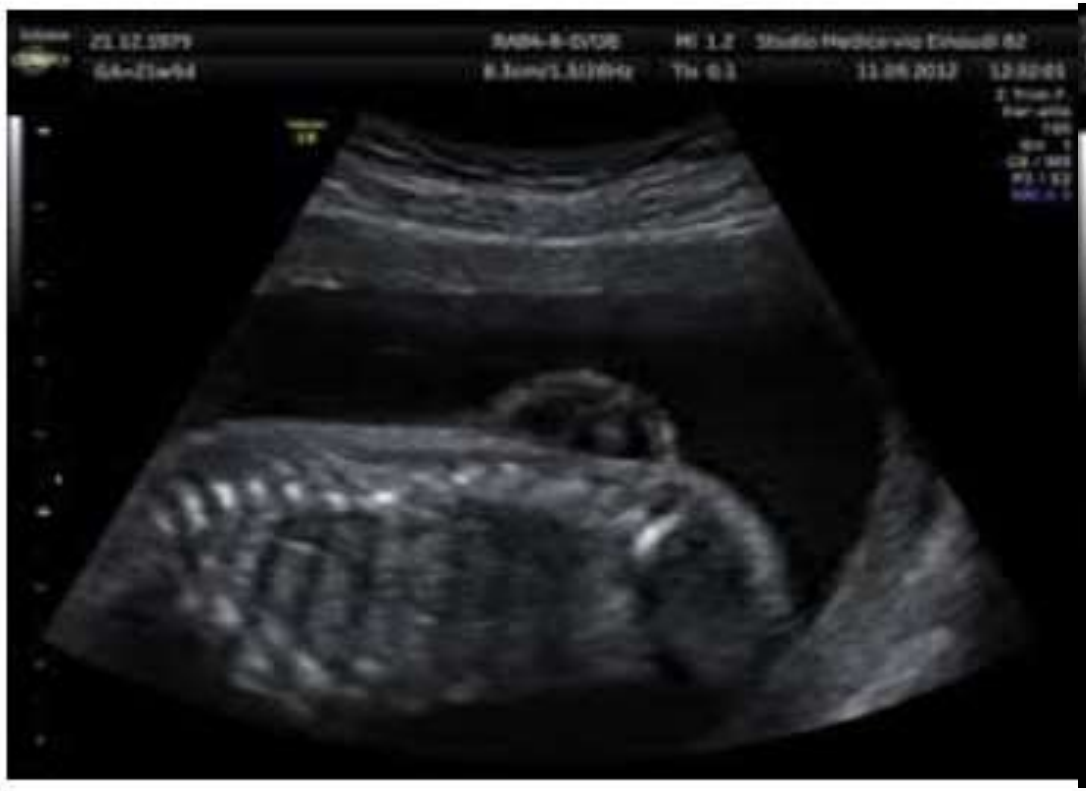
Part of infection: no increased risk.

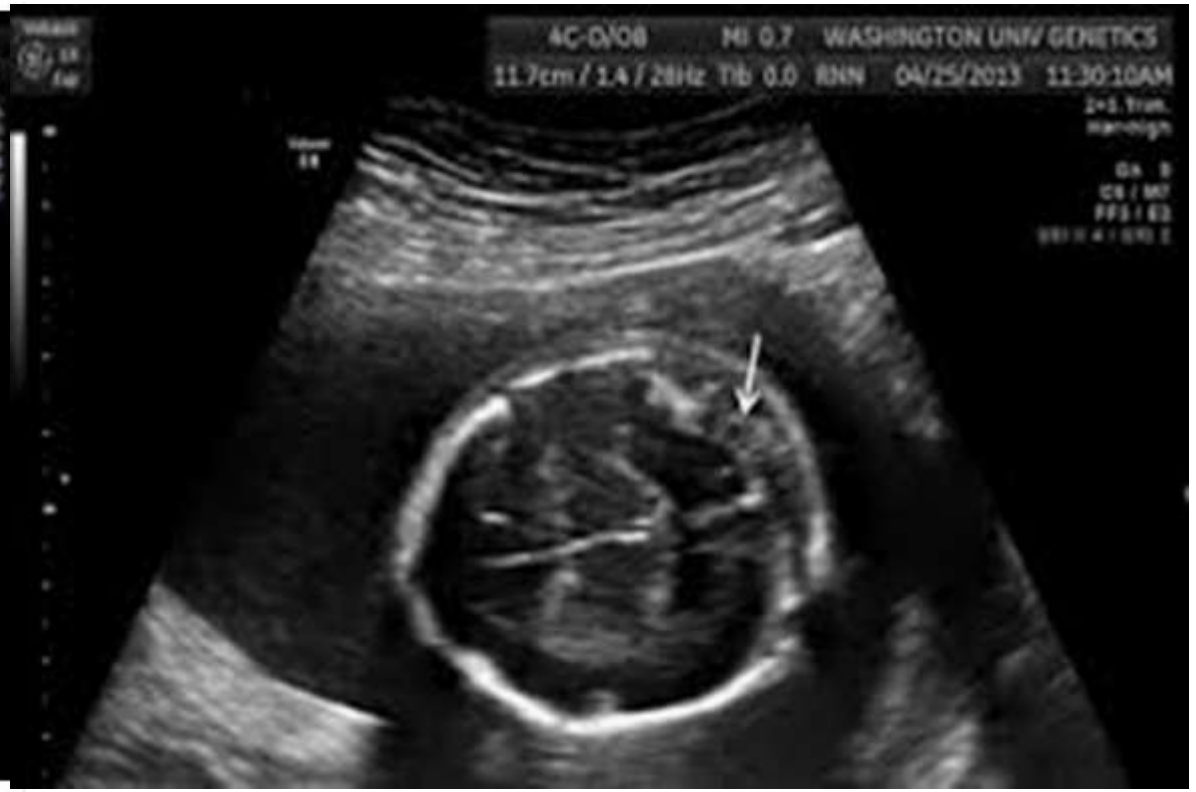
Part of trisomies: 1%.

X-linked hydrocephaly: 50% of males.

Associated with alloimmune thrombocytopenia and no treatment: 100%.

Open spina bifida





Open spina bifida

Prevalence:

1 in 1,000 fetuses at 12 weeks' gestation.

Site of open spina bifida: lumbosacral (65%), sacral (24%), thoracolumbar (10%), cervical (1%).

Associated abnormalities:

Chromosomal abnormalities (mainly trisomy 18), single mutant genes, and maternal diabetes mellitus or ingestion of antiepileptic drugs, are implicated in about 10% of the cases.

Risk of non-chromosomal syndromes is low.

Investigations:

Detailed ultrasound examination.

Fetal karyotyping in the presence of associated anomalies.

Follow up:

Follow-up scans every 4 weeks to monitor evolution of ventriculomegaly, possible kyphoscoliosis and development of talipes.

Prognosis:

The 5-year mortality rate is about 35%, with 20% dying during the first 12 months of life. About 25% of fetuses with spina bifida are stillborn

The surviving infants usually have paralysis in the lower limbs and double incontinence; despite the associated hydrocephalus requiring surgery, intelligence is often normal.

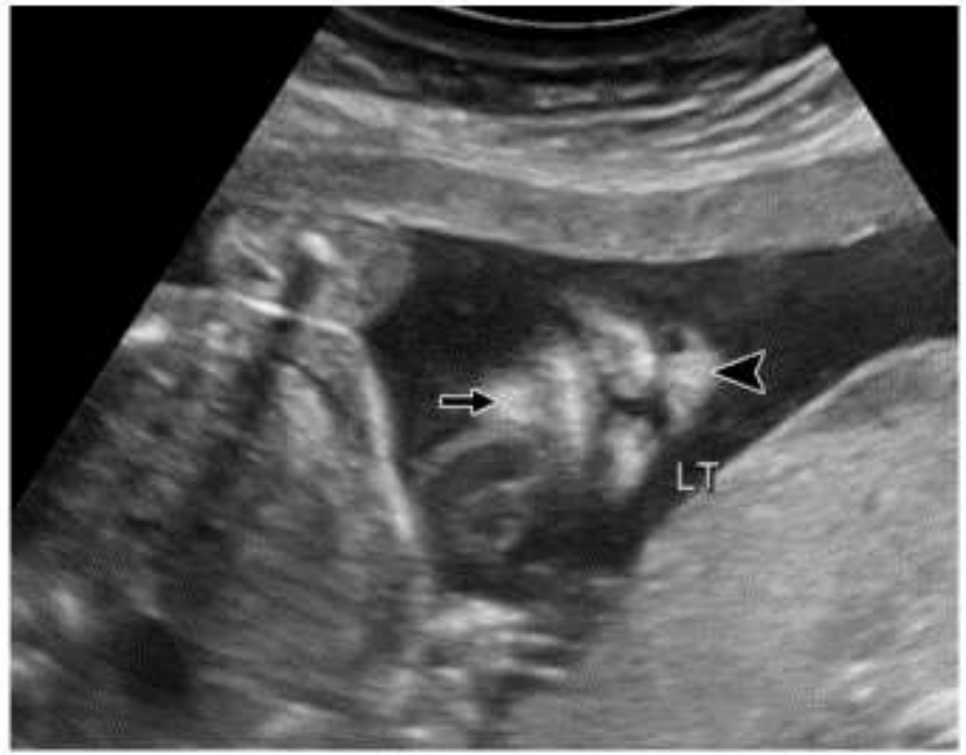
Recurrence:

Affected parent or one sibling: 5%.

Two affected siblings: 10%.

Supplementation of the maternal diet with folate (5 mg/day) for 3 months before and 2 months after conception reduces the risk of recurrence by about 75%.

Facial cleft



Facial cleft

Prevalence:

•1 in 700 births.
•More common in males than females and in Whites than Blacks.
•In 50% of cases, both the lip and palate are affected, in 25% only the lip and in 25% only the palate.
•Unilateral in 75% of cases (more common on the left side) and bilateral in 25%.

Associated abnormalities:

•Chromosomal abnormalities, mainly trisomies 13 and 18, are found in 1–2% of cases. Unilateral cleft lip is not associated with chromosomal abnormalities.
•Associated with any one of >400 syndromes in 30% of cases.

Investigations:

- Detailed ultrasound examination.
- Invasive testing for karyotyping and array.

Follow-up:

- Follow-up should be standard.
- Prenatal consultation with multidisciplinary team.

Delivery:

- Standard obstetric care and delivery.

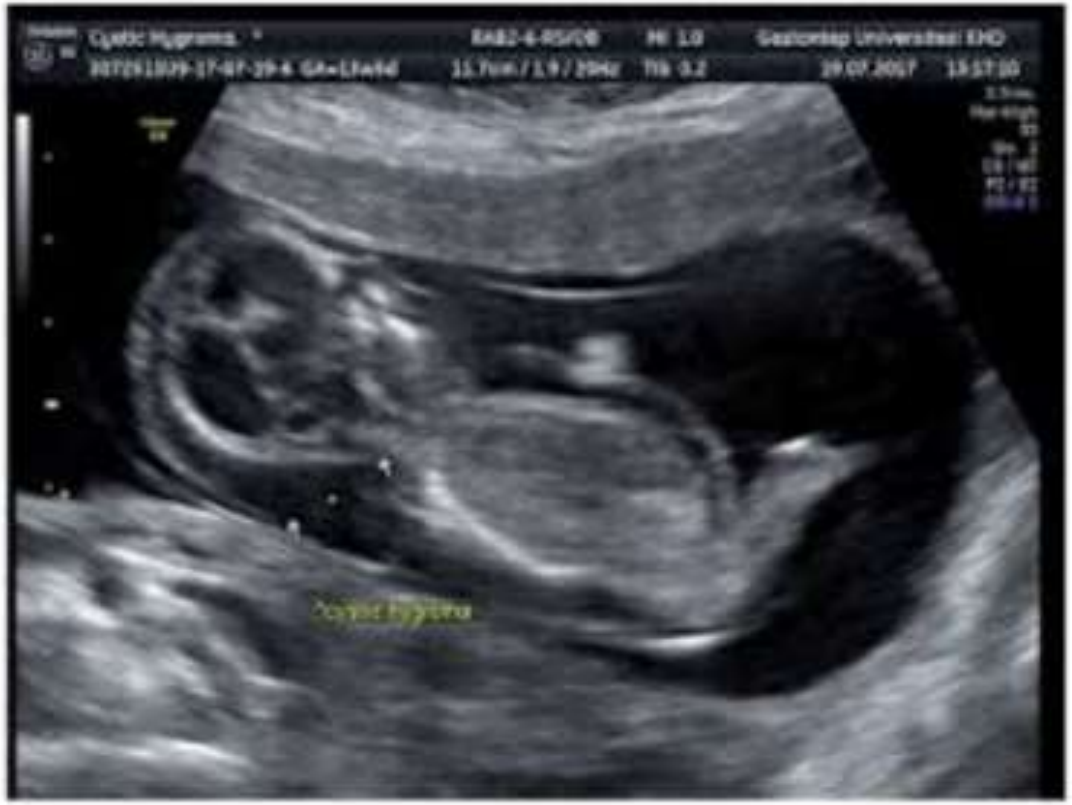
Prognosis:

- Isolated: Good prognosis and normal survival.
- Surgical repair is at 3-6 months of age.
- Long-term issues in children with cleft lip and palate include dental abnormalities, hearing and olfactory problems, midface hypoplasia, and psychological problems.

Recurrence:

- Isolated: 5% if one sibling or parent is affected and 10% if two siblings are affected.

Cystic hygroma



Cystic hygroma

Prevalence:

•1 in 800 pregnancies.

•1 in 8,000 live births.

Associated abnormalities:

•Chromosomal abnormalities, mainly Turner syndrome, are found in about 50% of cases.

•Genetic syndromes are found in about 40% of cases. The most common are **Noonan syndrome**

•Hydrops (in addition to cystic hygromas there is generalized edema, ascites, pericardial or pleural effusions) occurs in 60-80% of cases.

Investigations:

Detailed ultrasound examination, including echocardiography.

Invasive testing for karyotyping and array.

Follow-up:

Follow-up scans every 4 weeks to assess the evolution of the hygromas and development of hydrops.

Delivery:

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks, ealier if hydrops develops.

Method: cesarean section if there is hydrops or large cystic hygromas preventing flexion of the head.

Prognosis:

Fetal death: 90%.

In 10% of cases the fetal karyotype is normal, there are no other obvious defects and the hygromas resolve during pregnancy. In these cases the prognosis is good.

Recurrence:

Isolated or part of Turner syndrome: no increased risk of recurrence.

Part of autosomal recessive syndroms: 25%.

Diaphragmatic hernia

Prevalence:

1 in 4,000 births.

Associated abnormalities:

Chromosomal defects, mainly trisomies 18 or 13 .

Genetic syndromes are found in 10% of cases.

Defects in other systems, mainly craniofacial and cardiac, are found in 20% of cases.

Investigations:

Detailed ultrasound examination, including echocardiography.

Amniocentesis for karyotyping

Follow up:

Ultrasound scans every 4 weeks to monitor lung growth and amniotic fluid volume. Amniodrainage may be necessary if there is polyhydramnios and cervical shortening. Risk of fetal death about 5%.

Delivery:

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks. Earlier if there is evidence of poor growth or fetal hypoxia.

Method: induction of labor aiming for vaginal delivery.

Prognosis:

Part of trisomy 18: lethal.

Isolated: survival is <15% for severe disease, 50% for moderate disease and >90% for mild disease.

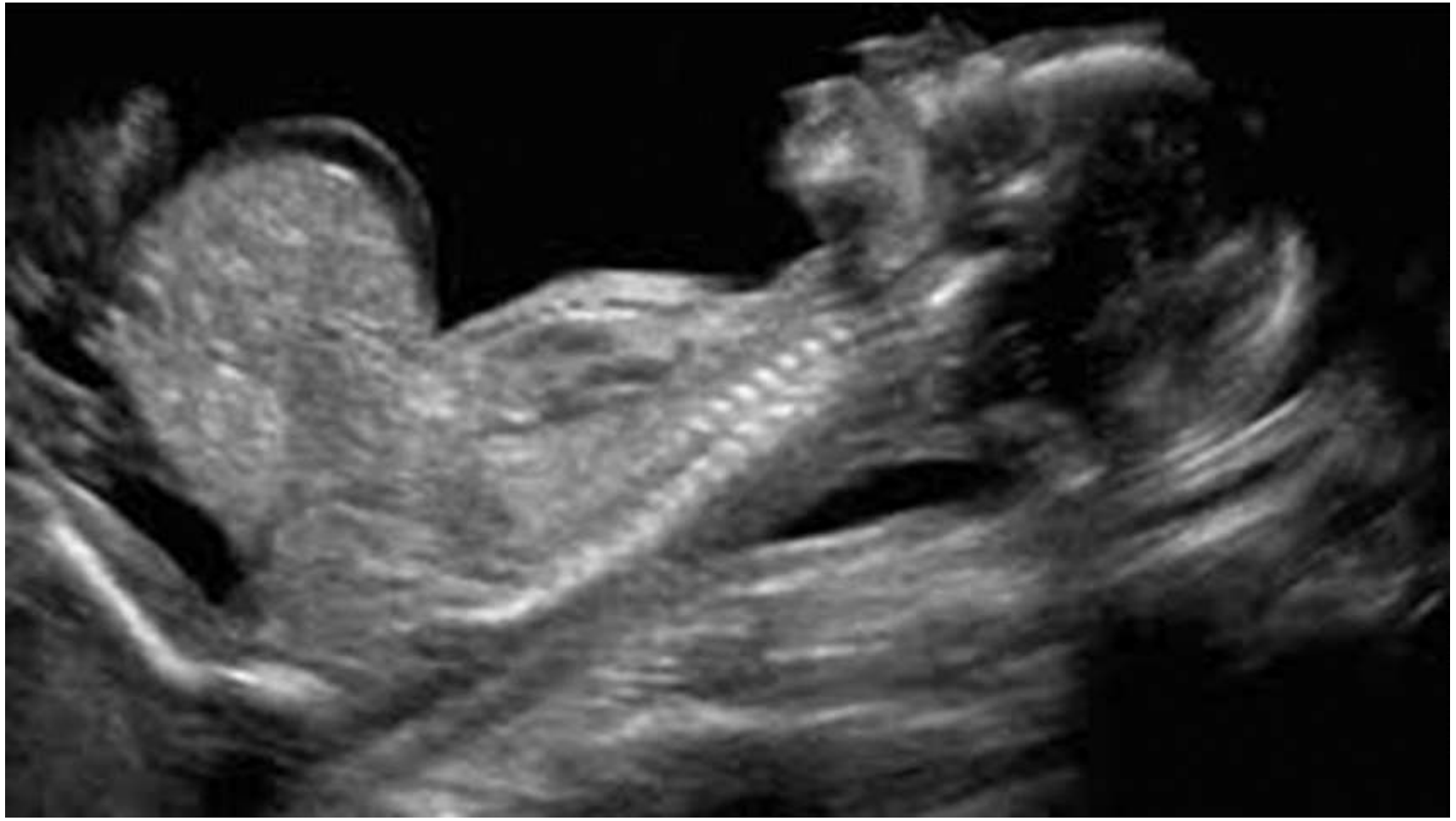
Recurrence:

Isolated: no increased risk.

Part of trisomies: 1%.

Part of syndromes: 25%.

omphalocele



Omphalocele

Prevalence:

Only bowel in the sac: 1 in 100 at 11 weeks' gestation, 1 in 800 at 12 weeks, 1 in 2,000 at 13 weeks.

Liver in the sac: 1 in 3,500 fetuses at 11 to 13 weeks.

Associated abnormalities:

Chromosomal defects, mainly trisomies 18 or 13, are found in 30-50% of cases.

Genetic syndromes,

Defects in other systems, mainly cardiac, are found in 30-50% of cases.

Investigations:

Detailed ultrasound examination, including echocardiography.

Invasive testing for karyotyping and molecular testing for Beckwith-Wiedemann syndrome.

Follow up:

Ultrasound scans every 4 weeks to monitor fetal growth and amniotic fluid. It is best to monitor growth through estimation of fetal weight by the Sieme formula which uses biparietal diameter, occipitofrontal diameter and femur length, rather than formulas using abdominal circumference.

Delivery:

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks. Earlier if there is evidence of poor growth or fetal hypoxia.

Method: induction of labor aiming for vaginal delivery. Cesarean section reserved for obstetric indications, such as breech presentation and for giant exomphalos (sac containing >75% of liver) to avoid rupture and hemorrhage.

Prognosis:

Isolated small / moderate exomphalos: survival >90%.

Isolated giant exomphalos: survival 80%.

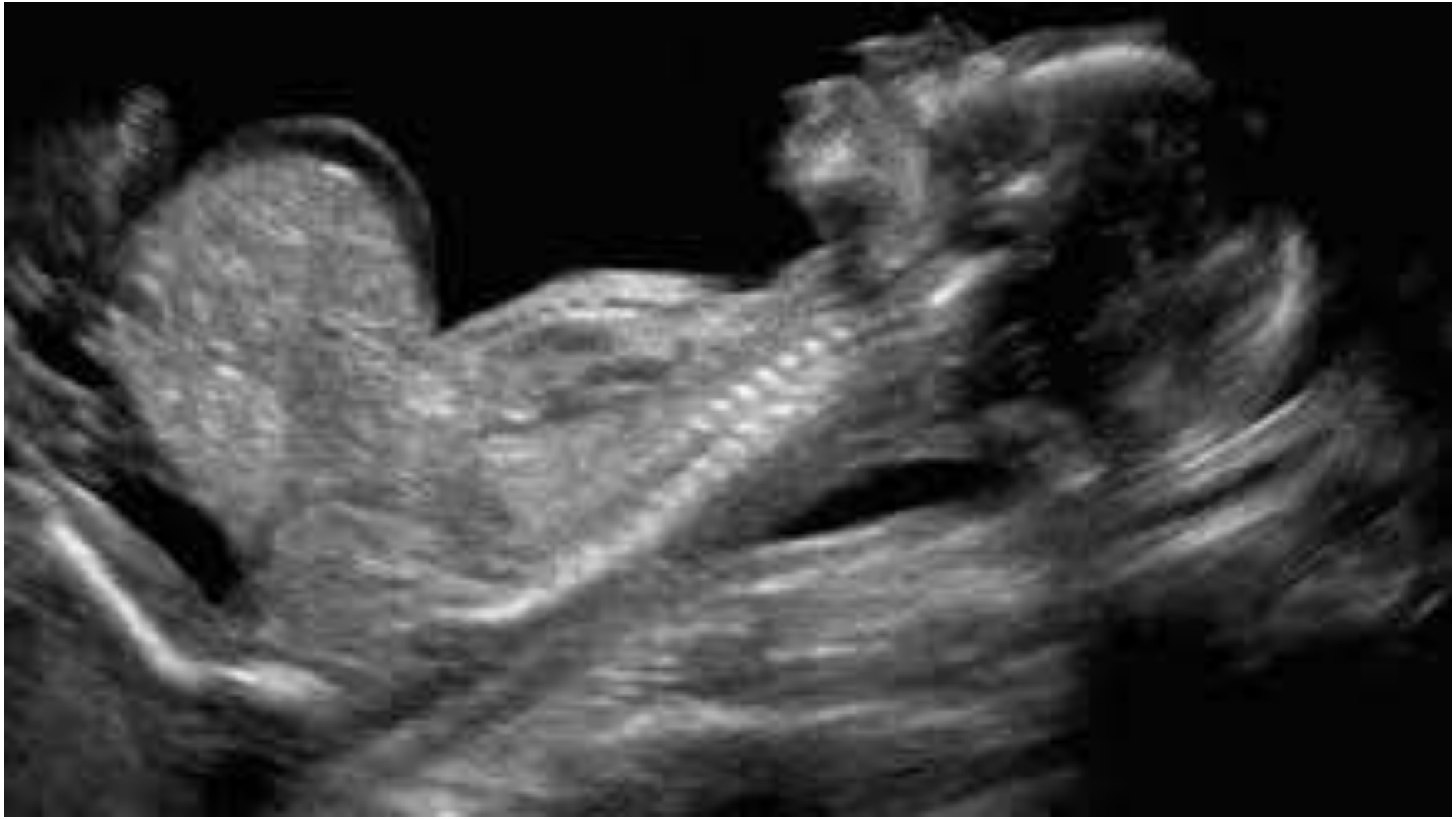
Other defect: depends on defect, e.g. trisomy 18 is lethal.

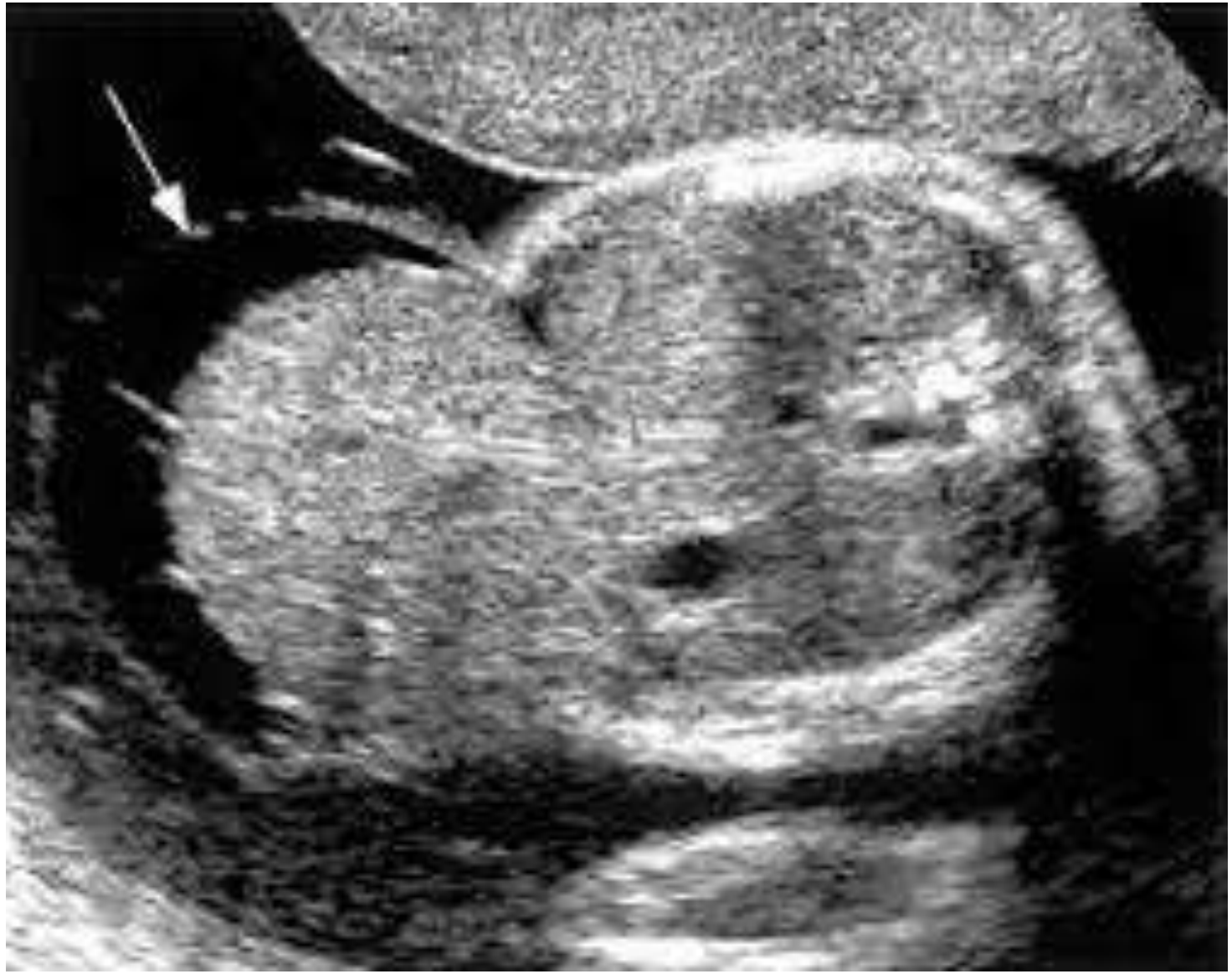
Recurrence:

Isolated: no increased risk.

Part of trisomies: 1%.

Part of Beckwith-Wiedemann syndrome: up to 50%.





Gastroschisis

Prevalence:

1 in 3,000 births.

Increased risk in young women and in those with cocaine abuse.

Associated abnormalities:

The incidence of chromosomal abnormalities and genetic syndromes is not increased.

Investigations:

Detailed ultrasound examination.

Associated complications:

Bowel atresias or obstruction secondary to volvulus and/or ischemia at the hernial orifice in about 10-30% of cases.

Fetal growth restriction in 30-60% of cases.

Spontaneous preterm birth in about 30% of cases.

Fetal death in 2-4% of cases.

Follow up:

Ultrasound scans every 4 weeks to monitor growth, amniotic fluid, fetal oxygenation (UA-PI, MCA-PI and DV-PI) and intra-abdominal bowel dilatation.

In fetuses with abdominal wall defects it is best to monitor growth through estimation of fetal weight by the Sieme formula which uses biparietal diameter, occipitofrontal diameter and femur length, rather than formulas using abdominal circumference.

Delivery:

Place: hospital with neonatal intensive care and pediatric surgery.

Time: 38 weeks. Earlier if there is evidence of poor growth, fetal hypoxia or dilatation of intrabdominal bowel (>20 mm).

Method: induction of labor aiming for vaginal delivery. There is no good evidence that cesarean section is beneficial.

Prognosis:

Survival: >90%

Recurrence: 3%.


<-- Omphalocele
Gastroschisis

Hepatic calcifications

Prevalence:

1 in 2,000 fetuses at 20 weeks' gestation.

Associated abnormalities:

The incidence of chromosomal abnormalities and genetic syndroms is not increased.

Parenchymal calcifications may be due to intrauterine infection.

Investigations:

Detailed ultrasound examination.

TORCH test for fetal infections.

Follow up:

In isolated cases follow-up should be standard.

Delivery:

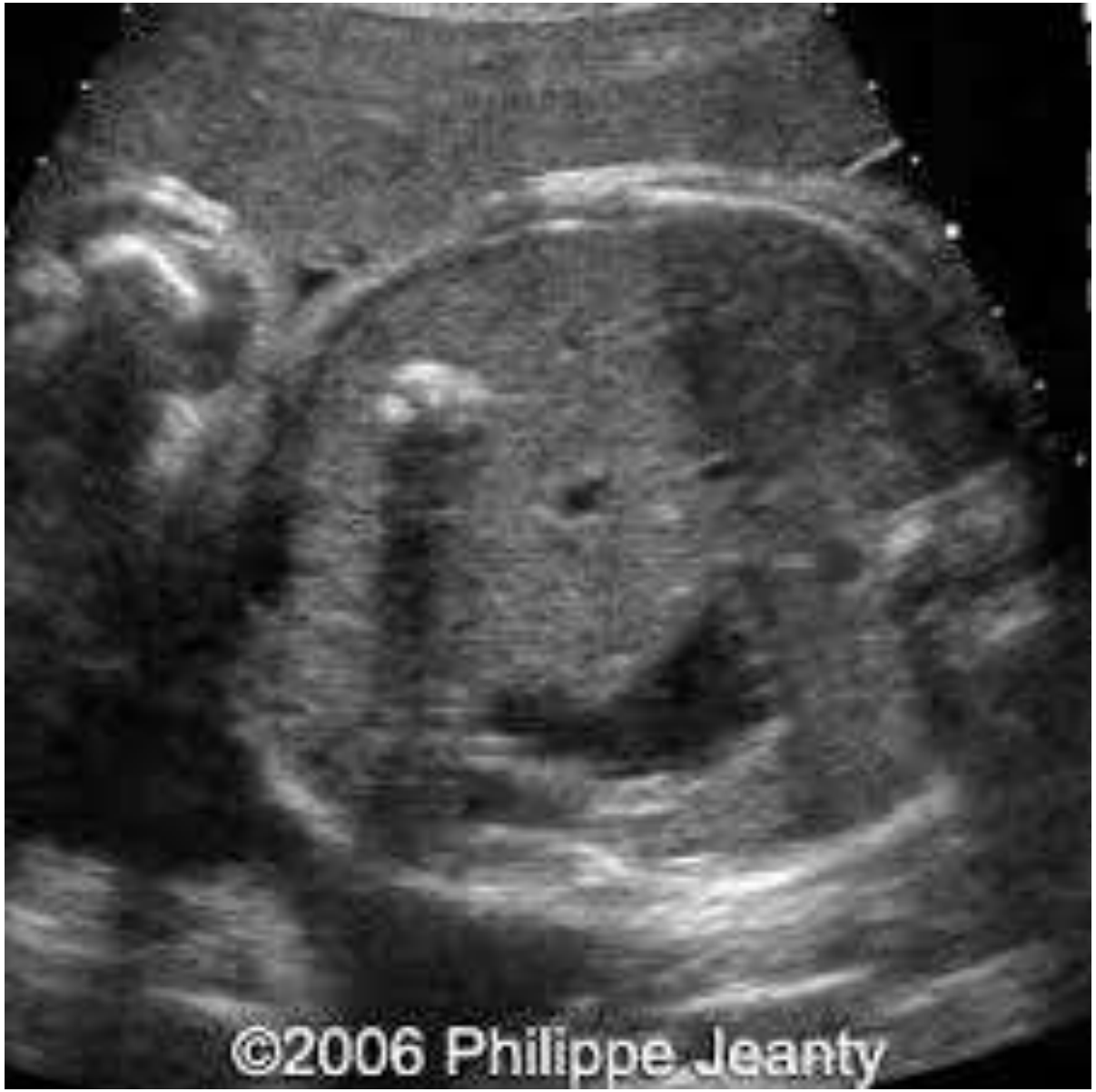
Standard obstetric care and delivery.

Prognosis:

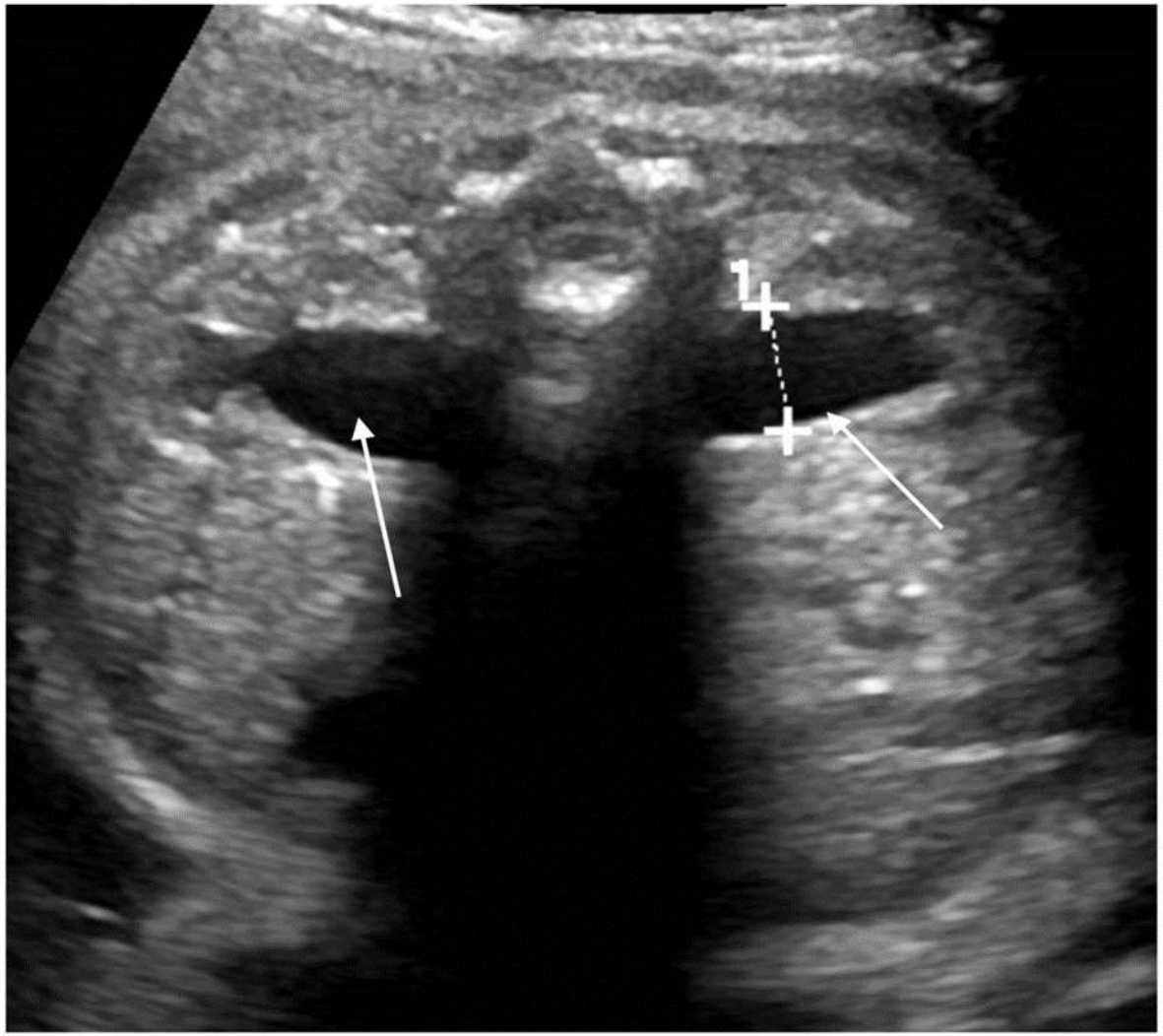
Good.

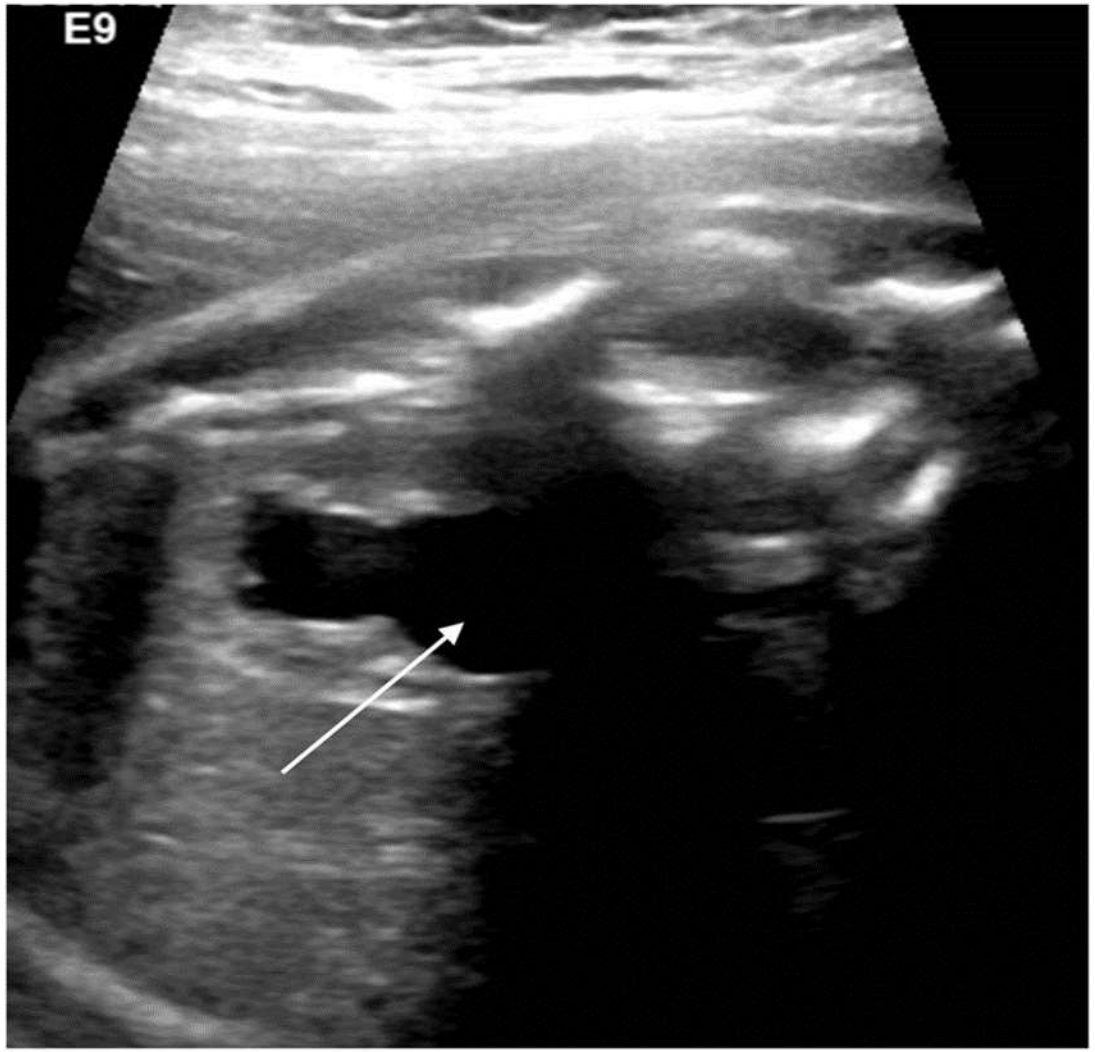
Recurrence:

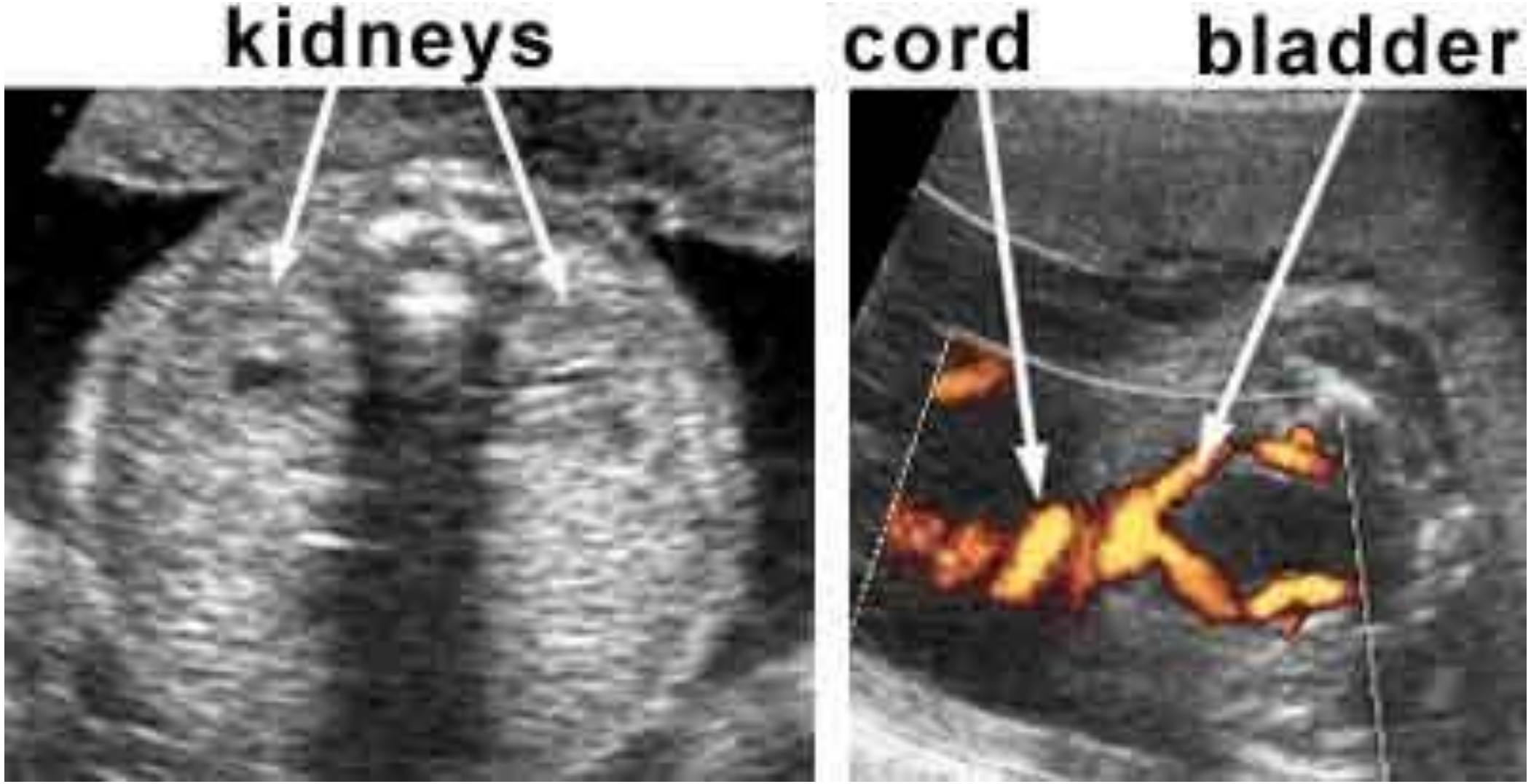
No increased risk of recurrence.


©2006 Philippe Jeanty

Hydronephrosis






kidneys
cord
bladder

Hydronephrosis

Prevalence:

1 in 500 births.

Mild (only renal pelvis): 4-7 mm in the 2nd trimester; 7-9 mm in the 3rd.

Moderate (pelvis and calyces): 8-10 mm in the 2nd trimester; 10-15 mm in the 3rd.

Severe (cortical thinning): >10 mm in the 2nd trimester; >15 mm in the 3rd. Associated abnormalities:

Chromosomal defects:

low risk in isolated forms.

Abnormalities of the contralateral kidney: multicystic kidney, ectopia, renal agenesis.

Investigations:

Detailed ultrasound examination (especially of the contralateral kidney).

Karyotyping should be offered only if other markers are present.

Follow up:

Ultrasound scans every 4 weeks to monitor the evolution of hydronephrosis and assess amniotic fluid volume.

Delivery:

Standard obstetric care and delivery.

Prognosis:

In the majority of cases, the condition remains stable or resolves in the neonatal period. Moderate hydronephrosis is usually progressive and in more than 50% of cases surgery is necessary during the first 2 years of life.

Recurrence:

Isolated: no increased risk of recurrence.

Clubfoot

Prevalence:

1 in 1,000 births.

Bilateral in 50% of cases.

Associated abnormalities:

In >50% of cases the condition is isolated.

Chromosomal abnormalities: common finding in trisomies 18 and 13.

Commonly associated with prolonged oligohydramnios, brain abnormalities, spina bifida, skeletal and neuromuscular disorders.

More than 250 genetic syndromes include clubfoot as one component.

Investigations:

Detailed ultrasound examination.

Non-isolated cases: invasive testing and array.

Follow up:

Isolated: standard follow-up.

Delivery:

Standard obstetric care and delivery.

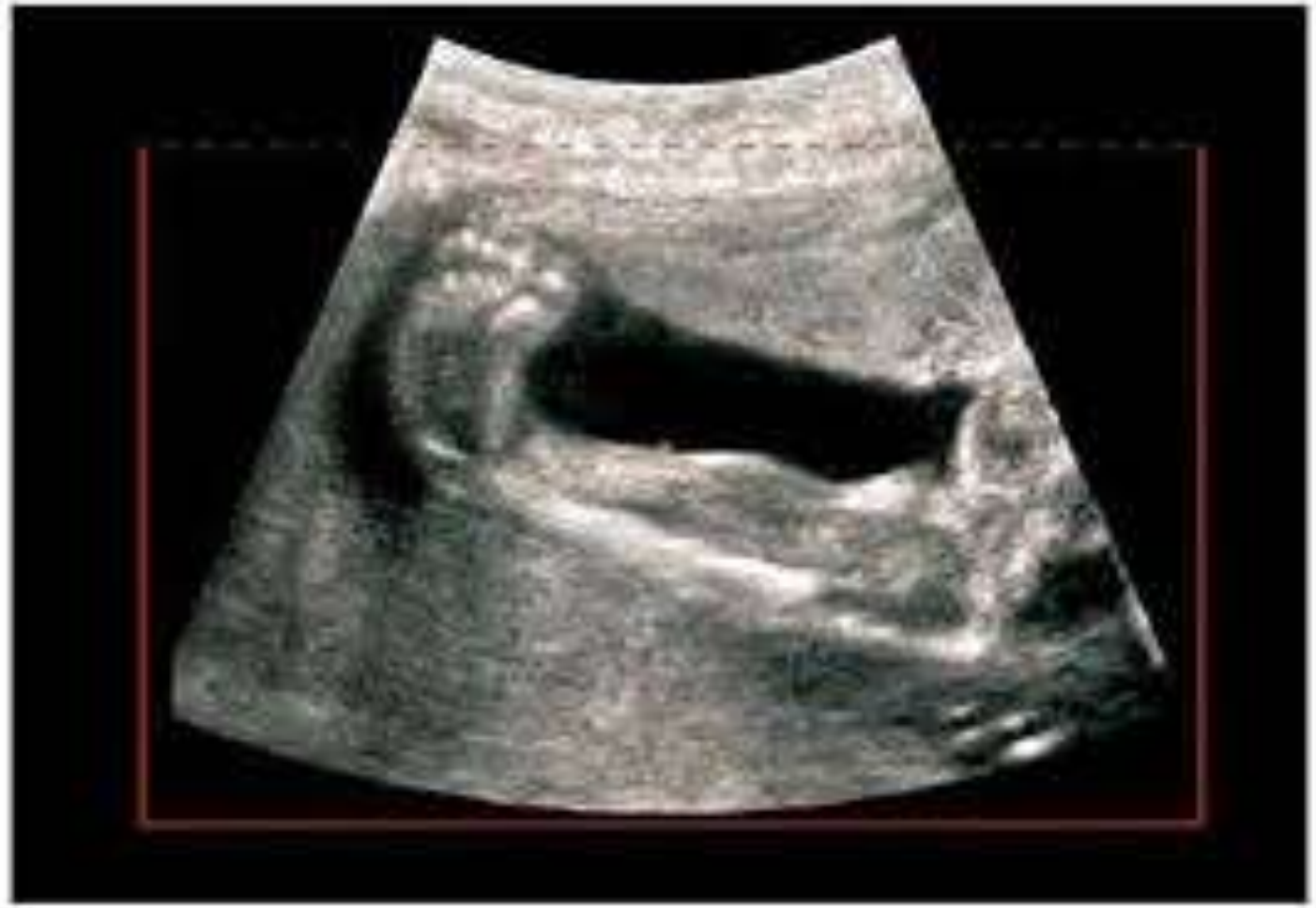
Prognosis:

Isolated: good prognosis.

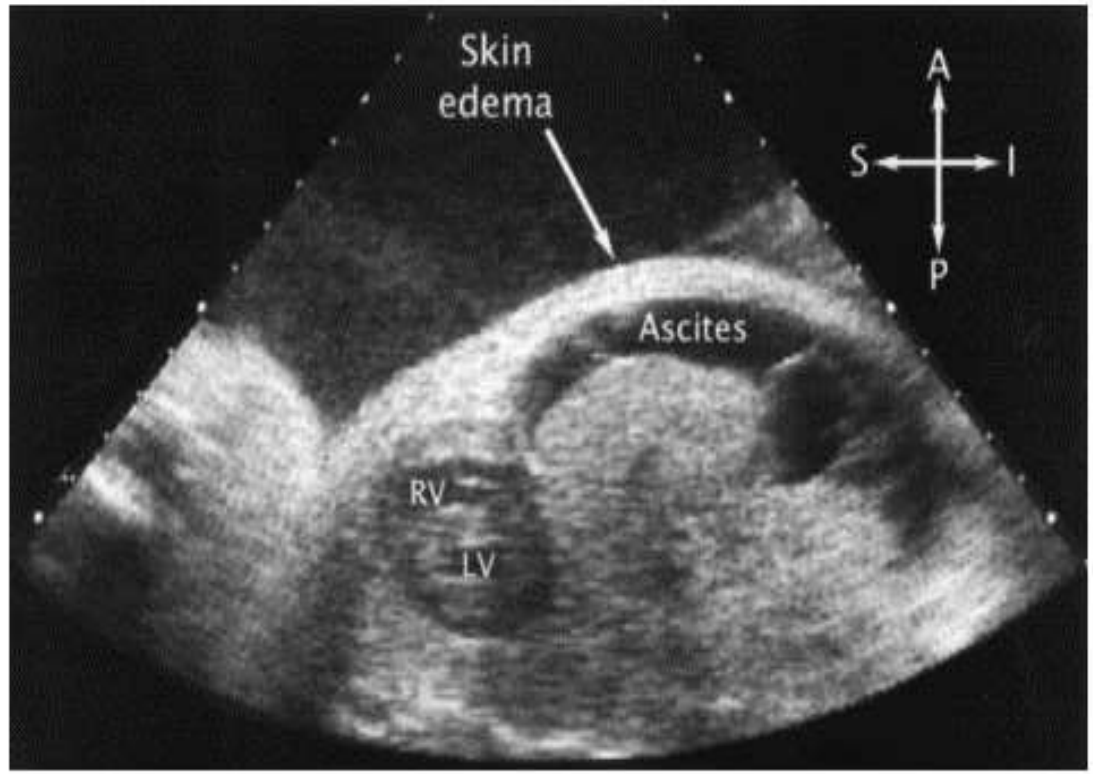
Recurrence:

One sibling with clubfoot: 3%.

Parent and one child with clubfoot: 25%.



Hydrops fetalis



Hydrops fetalis

Prevalence:

1 in 2,000 births.

Etiology:

Non-specific finding in a wide variety of fetal and maternal disorders, including hematological, chromosomal, cardiovascular, renal, pulmonary, gastrointestinal, hepatic and metabolic abnormalities, congenital infection, neoplasms, malformations of the placenta or umbilical cord and genetic syndromes.

Classically divided into:

- Immune: 10% of cases and it is due to maternal hemolytic antibodies.
- Non-immune: 90% of cases and it is due to all other etiologies.

Investigations:

In many instances, the underlying cause may be determined by maternal antibody and infection screening, fetal ultrasound scanning, including echocardiography, Doppler studies, fetal blood sampling and amniocentesis for karyotyping and array.

Often the abnormality remains unexplained even after expert post mortem examination.

Follow up:

Scans every 2-3 weeks to monitor the evolution of hydrops.

There is a risk of maternal morbidity due to the ‘mirror syndrome’ (combination of fetal hydrops with generalized fluid overload and a preeclamptic state in the mother).

Delivery:

Timing and method of delivery depend on the cause of hydrops.

Prognosis:

Depends on the cause of hydrops.

Progressive unexplained hydrops is often lethal before or soon after birth.

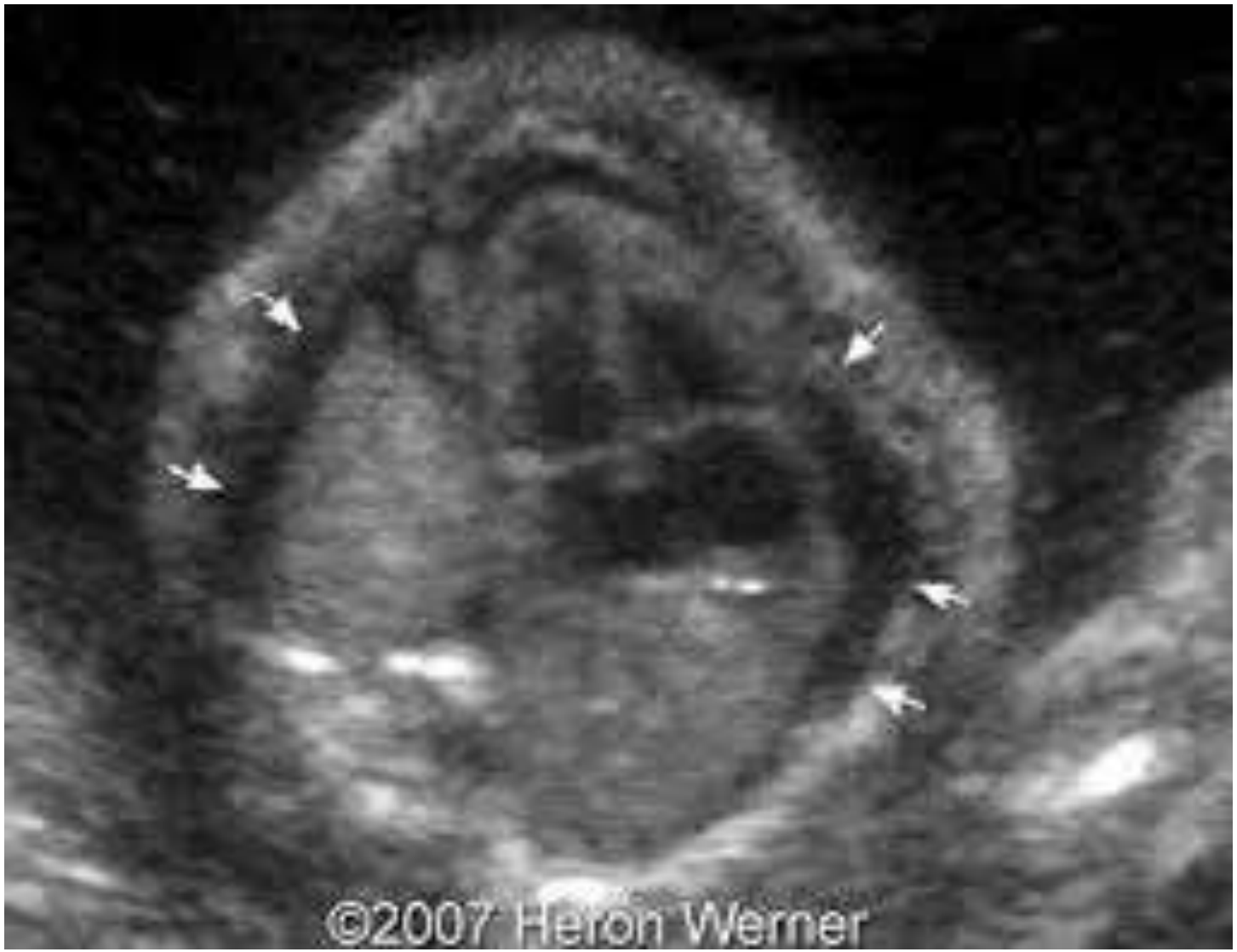
Recurrence:

Fetal defects, infection: no increased risk of recurrence.

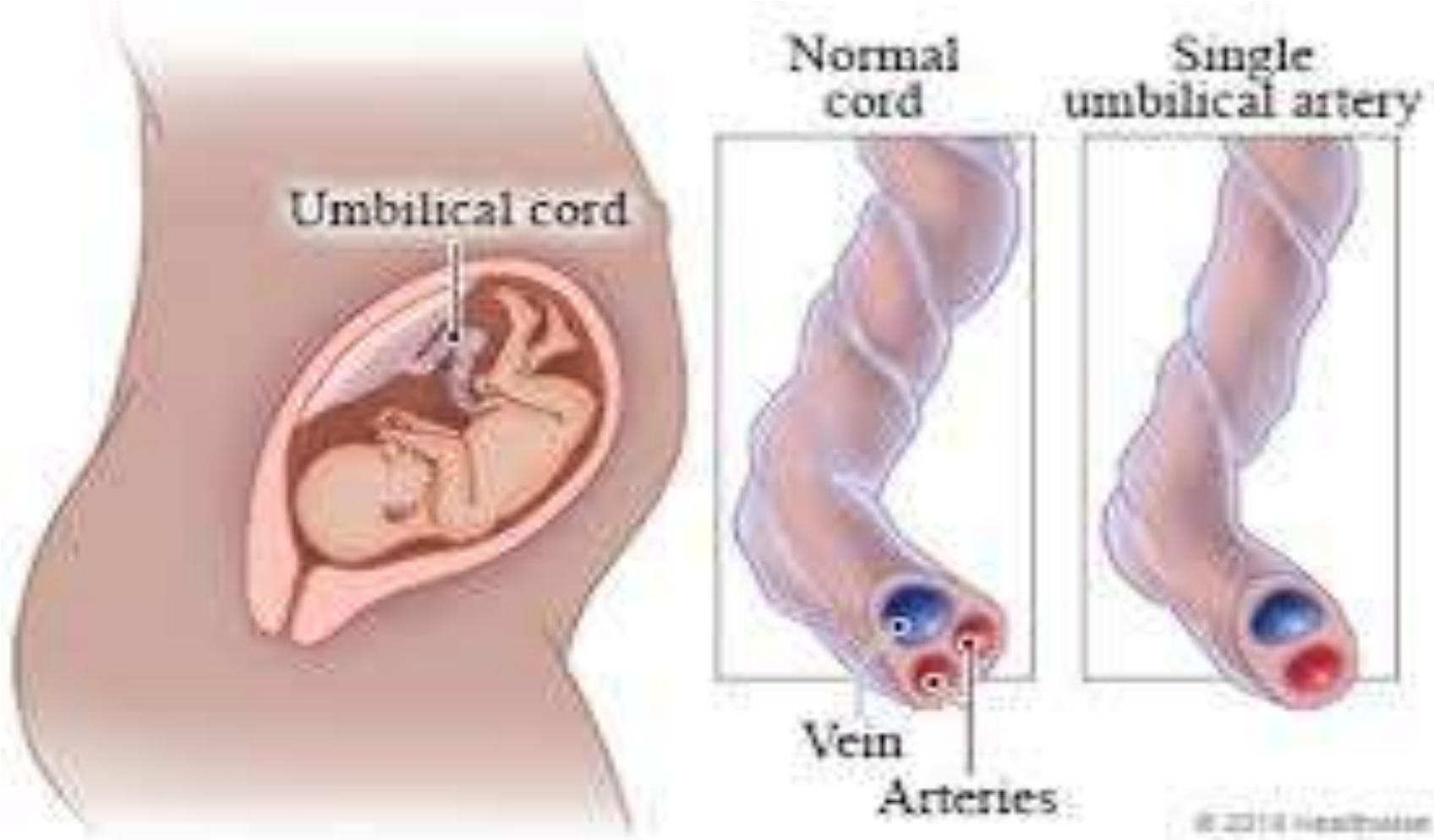
Part of trisomies: 1%.

Red blood cell isoimmunisation: high.

Metabolic disorders: 25%.


©2007 Heron Werner

Single umbilical artery



Single umbilical artery

Prevalence:

1 in 100 pregnancies.

Associated abnormalities:

Chromosomal abnormalities, mainly trisomy 18, 13 and triploidy, are found in 5% of cases.

Fetal growth restriction (<5th percentile) occurs in 10% of cases. Stillbirth, usually in association with growth restriction is twice as common as in the general population.

Abnormalities affecting cardiovascular, skeletal, gastrointestinal, genitourinary and central nervous systems, are found in 20% of cases.

Investigations:

Detailed ultrasound examination.

Fetal karyotyping in non-isolated cases.

Follow up:

Serial scans at 28, 32 and 36 weeks' gestation to assess fetal growth and well-being.

Delivery:

Standard obstetric care and delivery.

Prognosis:

In isolated cases the prognosis is normal.

Recurrence risk:

Isolated: no increased risk of recurrence.

Part of trisomies: 1%.